

<110> Samsung Electronics Co. Ltd

<120> A variant of HNF-1a gene having novel single nucleotide polymorphism and a variant protein encoded by the same

<130> YPL-0064

<140> 10/6663,857

<141> 2003-09-15

<160> 27

<170> KopatentIn 1.71

<210> 1

<211> 1896

<212> DNA

<213> Homo sapiens

<220>

<221> CDS

<222> (1)..(1893)

<223> amino acid sequence of HNF-1a

<400> 1

atg gtt tct aaa ctg agc cag ctg cag acg gag ctc ctg gcg gcc ctg	48
Met Val Ser Lys Leu Ser Gln Leu Gln Thr Glu Leu Leu Ala Ala Leu	
1 5 10 15	
ctc gag tca ggg ctg agc aaa gag gca ctg atc cag gca ctg ggt gag	96
Leu Glu Ser Gly Leu Ser Lys Glu Ala Leu Ile Gln Ala Leu Gly Glu	
20 25 30	
ccg ggg ccc tac ctc ctg gct gga gaa ggc ccc ctg gac aag ggg gag	144
Pro Gly Pro Tyr Leu Leu Ala Gly Glu Gly Pro Leu Asp Lys Gly Glu	
35 40 45	
tcc tgc ggc ggc ggt cga ggg gag ctg gct gag ctg ccc aat ggg ctg	192
Ser Cys Gly Gly Gly Arg Gly Glu Leu Ala Glu Leu Pro Asn Gly Leu	
50 55 60	
ggg gag act cgg ggc tcc gag gac gag acg gac gac gat ggg gaa gac	240
Gly Glu Thr Arg Gly Ser Glu Asp Glu Thr Asp Asp Asp Gly Glu Asp	
65 70 75 80	
ttc acg cca ccc atc ctc aaa gag ctg gag aac ctc agc cct gag gag	288
Phe Thr Pro Pro Ile Leu Lys Glu Leu Glu Asn Leu Ser Pro Glu Glu	
85 90 95	
gcg gcc cac cag aaa gcc gtg gtg gag acc ctt ctg cag gag gac ccg	336
Ala Ala His Gln Lys Ala Val Val Glu Thr Leu Leu Gln Glu Asp Pro	
100 105 110	
tgg cgt gtg gcg aag atg gtc aag tcc tac ctg cag cag cac aac atc	384
Trp Arg Val Ala Lys Met Val Lys Ser Tyr Leu Gln Gln His Asn Ile	
115 120 125	
cca cag cgg gag gtg gtc gat acc act ggc ctc aac cag tcc cac ctg	432

PX018432US_SEQ_1stOA_Amended.txt

Pro	Gln	Arg	Glu	Val	Val	Asp	Thr	Thr	Gly	Leu	Asn	Gln	Ser	His	Leu		
130						135				140							
tcc	caa	cac	ctc	aac	aag	ggc	act	ccc	atg	aag	acg	cag	aag	cgg	gcc		480
Ser	Gln	His	Leu	Asn	Lys	Gly	Thr	Pro	Met	Lys	Thr	Gln	Lys	Arg	Ala		
145				150					155					160			
gcc	ctg	tac	acc	tgg	tac	gtc	cgc	aag	cag	cga	gag	gtg	gcg	cag	cag		528
Ala	Leu	Tyr	Thr	Trp	Tyr	Val	Arg	Lys	Gln	Arg	Glu	Val	Ala	Gln	Gln		
				165					170					175			
ttc	acc	cat	gca	ggg	cag	gga	ggg	ctg	att	gaa	gag	ccc	aca	ggg	gat		576
Phe	Thr	His	Ala	Gly	Gln	Gly	Gly	Leu	Ile	Glu	Glu	Pro	Thr	Gly	Asp		
			180					185					190				
gag	cta	cca	acc	aag	aag	ggg	cgg	agg	aac	cgt	ttc	aag	tgg	ggc	cca		624
Glu	Leu	Pro	Thr	Lys	Lys	Gly	Arg	Arg	Asn	Arg	Phe	Lys	Trp	Gly	Pro		
		195					200					205					
gca	tcc	cag	cag	atc	ctg	ttc	cag	gcc	tat	gag	agg	cag	aag	aac	cct		672
Ala	Ser	Gln	Gln	Ile	Leu	Phe	Gln	Ala	Tyr	Glu	Arg	Gln	Lys	Asn	Pro		
	210					215					220						
agc	aag	gag	gag	cga	gag	acg	cta	gtg	gag	gag	tgc	aat	agg	gcg	gaa		720
Ser	Lys	Glu	Glu	Arg	Glu	Thr	Leu	Val	Glu	Glu	Cys	Asn	Arg	Ala	Glu		
225					230					235					240		
tgc	atc	cag	aga	ggg	gtg	tcc	cca	tca	cag	gca	cag	ggg	ctg	ggc	tcc		768
Cys	Ile	Gln	Arg	Gly	Val	Ser	Pro	Ser	Gln	Ala	Gln	Gly	Leu	Gly	Ser		
				245					250					255			
aac	ctc	gtc	acg	gag	gtg	cgt	gtc	tac	aac	tgg	ttt	gcc	aac	cgg	cgc		816
Asn	Leu	Val	Thr	Glu	Val	Arg	Val	Tyr	Asn	Trp	Phe	Ala	Asn	Arg	Arg		
			260					265					270				
aaa	gaa	gaa	gcc	ttc	cgg	cac	aag	ctg	gcc	atg	gac	acg	tac	agc	ggg		864
Lys	Glu	Glu	Ala	Phe	Arg	His	Lys	Leu	Ala	Met	Asp	Thr	Tyr	Ser	Gly		
		275					280					285					
ccc	ccc	cca	ggg	cca	ggc	ccg	gga	cct	gcg	ctg	ccc	gct	cac	agc	tcc		912
Pro	Pro	Pro	Gly	Pro	Gly	Pro	Gly	Pro	Ala	Leu	Pro	Ala	His	Ser	Ser		
		290				295					300						
cct	ggc	ctg	cct	cca	cct	gcc	ctc	tcc	ccc	agt	aag	gtc	cac	ggg	gtg		960
Pro	Gly	Leu	Pro	Pro	Pro	Ala	Leu	Ser	Pro	Ser	Lys	Val	His	Gly	Val		
305					310					315					320		
cgc	tat	gga	cag	cct	gcg	acc	agt	gag	act	gca	gaa	gta	ccc	tca	agc		1008
Arg	Tyr	Gly	Gln	Pro	Ala	Thr	Ser	Glu	Thr	Ala	Glu	Val	Pro	Ser	Ser		
				325					330					335			
agc	ggc	ggg	ccc	tta	gtg	aca	gtg	tct	aca	ccc	ctc	cac	caa	gtg	tcc		1056
Ser	Gly	Gly	Pro	Leu	Val	Thr	Val	Ser	Thr	Pro	Leu	His	Gln	Val	Ser		
			340					345					350				
ccc	acg	ggc	ctg	gag	ccc	agc	cac	agc	ctg	ctg	agt	aca	gaa	gcc	aag		1104
Pro	Thr	Gly	Leu	Glu	Pro	Ser	His	Ser	Leu	Leu	Ser	Thr	Glu	Ala	Lys		
		355					360					365					

PX018432US_SEQ_1stOA_Amended.txt

ctg gtc tca gca gct ggg ggc ccc ctc ccc cct gtc agc acc ctg aca Leu Val Ser Ala Ala Gly Gly Pro Leu Pro Pro Val Ser Thr Leu Thr 370 375 380	1152
gca ctg cac agc ttg gag cag aca tcc cca ggc ctc aac cag cag ccc Ala Leu His Ser Leu Glu Gln Thr Ser Pro Gly Leu Asn Gln Gln Pro 385 390 395 400	1200
cag aac ctc atc atg gcc tca ctt cct ggg gtc atg acc atc ggg cct Gln Asn Leu Ile Met Ala Ser Leu Pro Gly Val Met Thr Ile Gly Pro 405 410 415	1248
ggt gag cct gcc tcc ctg ggt cct acg ttc acc aac aca ggt gcc tcc Gly Glu Pro Ala Ser Leu Gly Pro Thr Phe Thr Asn Thr Gly Ala Ser 420 425 430	1296
acc ctg gtc atc ggc ctg gcc tcc acg cag gca cag agt gtg ccg gtc Thr Leu Val Ile Gly Leu Ala Ser Thr Gln Ala Gln Ser Val Pro Val 435 440 445	1344
atc aac agc atg ggc agc agc ctg acc acc ctg cag ccc gtc cag ttc Ile Asn Ser Met Gly Ser Ser Leu Thr Thr Leu Gln Pro Val Gln Phe 450 455 460	1392
tcc cag ccg ctg cac ccc tcc tac cag cag ccg ctc atg cca cct gtg Ser Gln Pro Leu His Pro Ser Tyr Gln Gln Pro Leu Met Pro Pro Val 465 470 475 480	1440
cag agc cat gtg acc cag aac ccc ttc atg gcc acc atg gct cag ctg Gln Ser His Val Thr Gln Asn Pro Phe Met Ala Thr Met Ala Gln Leu 485 490 495	1488
cag agc ccc cac gcc ctc tac agc cac aag ccc gag gtg gcc cag tac Gln Ser Pro His Ala Leu Tyr Ser His Lys Pro Glu Val Ala Gln Tyr 500 505 510	1536
acc cac acg ggc ctg ctc ccg cag act atg ctc atc acc gac acc acc Thr His Thr Gly Leu Leu Pro Gln Thr Met Leu Ile Thr Asp Thr Thr 515 520 525	1584
aac ctg agc gcc ctg gcc agc ctc acg ccc acc aag cag gtc ttc acc Asn Leu Ser Ala Leu Ala Ser Leu Thr Pro Thr Lys Gln Val Phe Thr 530 535 540	1632
tca gac act gag gcc tcc agt gag tcc ggg ctt cac acg ccg gca tct Ser Asp Thr Glu Ala Ser Ser Glu Ser Gly Leu His Thr Pro Ala Ser 545 550 555 560	1680
cag gcc acc acc ctc cac atc ccc agc cag gac cct gcc ggc atc cag Gln Ala Thr Thr Leu His Ile Pro Ser Gln Asp Pro Ala Gly Ile Gln 565 570 575	1728
cac ctg cag ccg gcc cac cgg ctc agc gcc agc ccc aca gtg tcc tcc His Leu Gln Pro Ala His Arg Leu Ser Ala Ser Pro Thr Val Ser Ser 580 585 590	1776
agc agc ctg gtg ctg tac cag agc tca gac tcc agc aat ggc cag agc Ser Ser Leu Val Leu Tyr Gln Ser Ser Asp Ser Ser Asn Gly Gln Ser 595 600 605	1824

PX018432US_SEQ_1stOA_Amended.txt

cac ctg ctg cca tcc aac cac agc gtc atc gag acc ttc atc tcc acc 1872
His Leu Leu Pro Ser Asn His Ser Val Ile Glu Thr Phe Ile Ser Thr
610 615 620

cag atg gcc tct tcc tcc cag taa 1896
Gln Met Ala Ser Ser Ser Gln
625 630

<210> 2
<211> 631
<212> PRT
<213> Homo sapiens

<400> 2
Met Val Ser Lys Leu Ser Gln Leu Gln Thr Glu Leu Leu Ala Ala Leu
1 5 10 15

Leu Glu Ser Gly Leu Ser Lys Glu Ala Leu Ile Gln Ala Leu Gly Glu
20 25 30

Pro Gly Pro Tyr Leu Leu Ala Gly Glu Gly Pro Leu Asp Lys Gly Glu
35 40 45

Ser Cys Gly Gly Gly Arg Gly Glu Leu Ala Glu Leu Pro Asn Gly Leu
50 55 60

Gly Glu Thr Arg Gly Ser Glu Asp Glu Thr Asp Asp Asp Gly Glu Asp
65 70 75 80

Phe Thr Pro Pro Ile Leu Lys Glu Leu Glu Asn Leu Ser Pro Glu Glu
85 90 95

Ala Ala His Gln Lys Ala Val Val Glu Thr Leu Leu Gln Glu Asp Pro
100 105 110

Trp Arg Val Ala Lys Met Val Lys Ser Tyr Leu Gln Gln His Asn Ile
115 120 125

Pro Gln Arg Glu Val Val Asp Thr Thr Gly Leu Asn Gln Ser His Leu
130 135 140

Ser Gln His Leu Asn Lys Gly Thr Pro Met Lys Thr Gln Lys Arg Ala
145 150 155 160

Ala Leu Tyr Thr Trp Tyr Val Arg Lys Gln Arg Glu Val Ala Gln Gln
165 170 175

Phe Thr His Ala Gly Gln Gly Gly Leu Ile Glu Glu Pro Thr Gly Asp
180 185 190

Glu Leu Pro Thr Lys Lys Gly Arg Arg Asn Arg Phe Lys Trp Gly Pro
195 200 205

Ala Ser Gln Gln Ile Leu Phe Gln Ala Tyr Glu Arg Gln Lys Asn Pro
210 215 220

Ser Lys Glu Glu Arg Glu Thr Leu Val Glu Glu Cys Asn Arg Ala Glu

PX018432US_SEQ_1stOA_Amended.txt

225		230		235		240									
Cys	Ile	Gln	Arg	Gly	Val	Ser	Pro	Ser	Gln	Ala	Gln	Gly	Leu	Gly	Ser
				245					250					255	
Asn	Leu	Val	Thr	Glu	Val	Arg	Val	Tyr	Asn	Trp	Phe	Ala	Asn	Arg	Arg
			260					265					270		
Lys	Glu	Glu	Ala	Phe	Arg	His	Lys	Leu	Ala	Met	Asp	Thr	Tyr	Ser	Gly
		275						280				285			
Pro	Pro	Pro	Gly	Pro	Gly	Pro	Gly	Pro	Ala	Leu	Pro	Ala	His	Ser	Ser
	290					295					300				
Pro	Gly	Leu	Pro	Pro	Pro	Ala	Leu	Ser	Pro	Ser	Lys	Val	His	Gly	Val
305					310					315					320
Arg	Tyr	Gly	Gln	Pro	Ala	Thr	Ser	Glu	Thr	Ala	Glu	Val	Pro	Ser	Ser
				325					330					335	
Ser	Gly	Gly	Pro	Leu	Val	Thr	Val	Ser	Thr	Pro	Leu	His	Gln	Val	Ser
			340					345					350		
Pro	Thr	Gly	Leu	Glu	Pro	Ser	His	Ser	Leu	Leu	Ser	Thr	Glu	Ala	Lys
		355						360				365			
Leu	Val	Ser	Ala	Ala	Gly	Gly	Pro	Leu	Pro	Pro	Val	Ser	Thr	Leu	Thr
	370					375					380				
Ala	Leu	His	Ser	Leu	Glu	Gln	Thr	Ser	Pro	Gly	Leu	Asn	Gln	Gln	Pro
385					390					395					400
Gln	Asn	Leu	Ile	Met	Ala	Ser	Leu	Pro	Gly	Val	Met	Thr	Ile	Gly	Pro
				405					410					415	
Gly	Glu	Pro	Ala	Ser	Leu	Gly	Pro	Thr	Phe	Thr	Asn	Thr	Gly	Ala	Ser
			420					425					430		
Thr	Leu	Val	Ile	Gly	Leu	Ala	Ser	Thr	Gln	Ala	Gln	Ser	Val	Pro	Val
		435						440				445			
Ile	Asn	Ser	Met	Gly	Ser	Ser	Leu	Thr	Thr	Leu	Gln	Pro	Val	Gln	Phe
	450					455					460				
Ser	Gln	Pro	Leu	His	Pro	Ser	Tyr	Gln	Gln	Pro	Leu	Met	Pro	Pro	Val
465					470					475					480
Gln	Ser	His	Val	Thr	Gln	Asn	Pro	Phe	Met	Ala	Thr	Met	Ala	Gln	Leu
				485					490					495	
Gln	Ser	Pro	His	Ala	Leu	Tyr	Ser	His	Lys	Pro	Glu	Val	Ala	Gln	Tyr
			500					505					510		
Thr	His	Thr	Gly	Leu	Leu	Pro	Gln	Thr	Met	Leu	Ile	Thr	Asp	Thr	Thr
		515						520				525			
Asn	Leu	Ser	Ala	Leu	Ala	Ser	Leu	Thr	Pro	Thr	Lys	Gln	Val	Phe	Thr
	530					535					540				

PX018432US_SEQ_1stOA_Amended.txt

Ser Asp Thr Glu Ala Ser Ser Glu Ser Gly Leu His Thr Pro Ala Ser
545 550 555 560

Gln Ala Thr Thr Leu His Ile Pro Ser Gln Asp Pro Ala Gly Ile Gln
565 570 575

His Leu Gln Pro Ala His Arg Leu Ser Ala Ser Pro Thr Val Ser Ser
580 585 590

Ser Ser Leu Val Leu Tyr Gln Ser Ser Asp Ser Ser Asn Gly Gln Ser
595 600 605

His Leu Leu Pro Ser Asn His Ser Val Ile Glu Thr Phe Ile Ser Thr
610 615 620

Gln Met Ala Ser Ser Ser Gln
625 630

<210> 3
<211> 93
<212> DNA
<213> Homo sapiens

<400> 3
gtaaggtcca ggctgtctgg ccctcccttg gctgtgaca gagcccctca cccccacatc 60
ccccgggctc aggaggctgc tctgctcccc cag 93

<210> 4
<211> 41
<212> DNA
<213> Artificial Sequence

<220>
<223> sense primer for amplifying promoter of MODY3 gene

<400> 4
taatacgact cactataggg tggccgtgag catcctctgc c 41

<210> 5
<211> 39
<212> DNA
<213> Artificial Sequence

<220>
<223> antisense primer for amplifying promoter of MODY3 gene

<400> 5
gtaaccctca ctaaaggac gtgggttgcg tttgcctgc 39

<210> 6
<211> 40
<212> DNA

PX018432US_SEQ_1stOA_Amended.txt

<213> Artificial Sequence

<220>

<223> sense primer for amplifying exon 1 of MODY3 gene

<400> 6
taatacgact cactataggg cgtggccctg tggcagccga 40

<210> 7
<211> 40
<212> DNA
<213> Artificial Sequence

<220>

<223> antisense primer for amplifying exon 1 of MODY3 gene

<400> 7
gtaaccctca ctaaaggag ggctcgtag gagctgaggg 40

<210> 8
<211> 42
<212> DNA
<213> Artificial Sequence

<220>

<223> sense primer for amplifying exon 2 of MODY3 gene

<400> 8
taatacgact cactataggg cccttgctga gcagatcccg tc 42

<210> 9
<211> 40
<212> DNA
<213> Artificial Sequence

<220>

<223> antisense primer for amplifying exon 2 of MODY3 gene

<400> 9
gtaaccctca ctaaaggag ggatggtgaa gcttcagcc 40

<210> 10
<211> 40
<212> DNA
<213> Artificial Sequence

<220>

<223> sense primer for amplifying exon 3 of MODY3 gene

<400> 10

PX018432US_SEQ_1stOA_Amended.txt

taatacgact cactataggg gcaaggtcag gggaatggac 40

<210> 11
 <211> 42
 <212> DNA
 <213> Artificial Sequence

<220>
 <223> antisense primer for amplifying exon 3 of MODY3 gene

<400> 11
 gtaaccctca ctaaaggac gccgttgtag ctattgcact cc 42

<210> 12
 <211> 43
 <212> DNA
 <213> Artificial Sequence

<220>
 <223> sense primer for amplifying exon 4 of MODY3 gene

<400> 12
 taatacgact cactataggg ggctcatggg tggctatttc tgc 43

<210> 13
 <211> 42
 <212> DNA
 <213> Artificial Sequence

<220>
 <223> antisense primer for amplifying exon 4 of MODY3 gene

<400> 13
 gtaaccctca ctaaaggac gtgtcccttg tccccacata cc 42

<210> 14
 <211> 42
 <212> DNA
 <213> Artificial Sequence

<220>
 <223> sense primer for amplifying exon 5 of MODY3 gene

<400> 14
 taatacgact cactataggg tgctgaggca ggacactgct tc 42

<210> 15
 <211> 42
 <212> DNA
 <213> Artificial Sequence

<220>
 <223> antisense primer for amplifying exon 5 of MODY3 gene

<400> 15
 gtaaccctca ctaaagggat acaagcaagg acactcacca gc 42

<210> 16
 <211> 41
 <212> DNA
 <213> Artificial Sequence

<220>
 <223> sense primer for amplifying exon 6 of MODY3 gene

<400> 16
 taatacgact cactataggg cccggacaca gcttggttc c 41

<210> 17
 <211> 42
 <212> DNA
 <213> Artificial Sequence

<220>
 <223> antisense primer for amplifying exon 6 of MODY3 gene

<400> 17
 gtaaccctca ctaaaggga tccccaccag cttaccgatg ac 42

<210> 18
 <211> 40
 <212> DNA
 <213> Artificial Sequence

<220>
 <223> sense primer for amplifying exon 7 of MODY3 gene

<400> 18
 taatacgact cactataggg caggcctggc ctccacgcag 40

<210> 19
 <211> 40
 <212> DNA
 <213> Artificial Sequence

<220>
 <223> antisense primer for amplifying exon 7 of MODY3 gene

<400> 19
 gtaaccctca ctaaaggag gggctctgca gctgagccat 40

<210> 20
 <211> 41
 <212> DNA
 <213> Artificial Sequence

 <220>
 <223> sense primer for amplifying exon 8 and 9 of MODY3 gene

 <400> 20
 taatacgact cactataggg ggcccagtac acccacacgg g 41

 <210> 21
 <211> 40
 <212> DNA
 <213> Artificial Sequence

 <220>
 <223> antisense primer for amplifying exon 8 and 9 of MODY3 gene

 <400> 21
 gtaaccctca ctaaaggagg ggcagggaca gtaaggagg 40

 <210> 22
 <211> 41
 <212> DNA
 <213> Artificial Sequence

 <220>
 <223> sense primer for amplifying exon 10 of MODY3 gene

 <400> 22
 taatacgact cactataggg gccttgtttg cctctgcagt g 41

 <210> 23
 <211> 41
 <212> DNA
 <213> Artificial Sequence

 <220>
 <223> antisense primer for amplifying exon 10 of MODY3 gene

 <400> 23
 gtaaccctca ctaaaggagg gccatctggg tggagatgaa g 41

 <210> 24
 <211> 20
 <212> DNA
 <213> Artificial Sequence

PX018432US_SEQ_1stOA_Amended.txt

<220>

<223> T7 promoter sequence

<400> 24

taatacgact cactataggg

20

<210> 25

<211> 19

<212> DNA

<213> Artificial Sequence

<220>

<223> T3 promoter sequence

<400> 25

gtaaccctca ctaaagga

19

<210> 26

<211> 540

<212> DNA

<213> Homo sapiens

<400> 26

gaggcctggg actagggctg tcaggcacgt ttgccacgtc tgcccctctc tcccctgcgg 60

ccagccctct acagccacaa gcccgaggtg gccagtaga cccacacggg cctgctcccg 120

cagactatgc tcataccga caccaccaac ctgagcgccc tggccagcct cagcccacc 180

aagcaggtaa ggtccaggcc tgctggcct cctcggcct gtgacagagc ccctcacc 240

cacatcccc gggctcagga ggctgctctg ctccccagg tcttcacctc agacactgag 300

gcctccagt agtccgggt tcacacgccc gcattcagg ccaccaccct ccacatcccc 360

agccaggacc ctgccggcat ccagcacctg cagccggccc accggctcag cgccagcccc 420

acaggtgaga ggccctggct ccacccctc ccttactgtc cctgccccct tccatgttgg 480

tcccaccct tctgttgctg tccgtcactg tggggctgtg catgcagcag gcctagggt 540

540

<210> 27

<211> 540

<212> DNA

<213> Homo sapiens

<400> 27

gaggcctggg actagggctg tcaggcacgt ttgccacgtc tgcccctctc tcccctgcgg 60

ccagccctct acagccacaa gcccgaggtg gccagtaga cccacacggg cctgctcccg 120

cagactatgc tcataccga caccaccaac ctgagcgccc tggccagcct cagcccacc 180

PX018432US_SEQ_1stOA_Amended.txt

aagcaggtaa ggtccaggcc tgctggccct cccttggcct gtgacagagc ccctcacccc	240
cacatcccc gggtcagga ggctgctctg ctccccagg tcttcacctc agacactgag	300
gcctccagtg agtcgggct tcacacgccg gcctctcagg ccaccacct ccacgtcccc	360
agccaggacc ctgccggcat ccagcacctg cagccggccc accggctcag cggcagcccc	420
acaggtgaga ggccctggct ccaccccctc ccttactgtc cctgccccct tccatgttgg	480
tcccaccct tctgttgctg tccgtcactg tggggctgtg catgcagcag gcctagggct	540
	540